

# The BRCA2 Gene

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- ⌘ BRCA2 is an autosomal dominant gene.
- ⌘ BRCA2 accounts for approximately 3-4% of all breast cancers.

# Clinical Significance

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- ⌘ Women with BRCA2 have a 50-85% chance of developing breast cancer, and a 15-25% chance of developing ovarian cancer.
- ⌘ Men with BRCA2 have a increased chance of developing male breast cancer.
- ⌘ BRCA2 is associated with other cancers, including prostate and pancreatic cancer.

# Possible Surveillance

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- ⌘ Women who know they have the BRCA2 gene can practice careful surveillance, including yearly mammography and trans-vaginal ultrasound.
- ⌘ It is unclear whether increased surveillance improves survival.

# Interventions for Women

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- ⌘ Mastectomy and tamoxifen may reduce the risk of breast cancer.
- ⌘ Oophorectomy may reduce the risk of ovarian cancer.
- ⌘ Oral contraceptives may reduce the risk of ovarian cancer, but may increase the risk of breast cancer.

# Family Studies

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- ⌘ The clinical genetics branch of NCI has been conducting a number of family studies over the past 15 years.
- ⌘ All family members who have participated in previous studies are offered genetic testing for the gene that has been identified in their family.

# Study Testing

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- ⌘ The researchers promise not to share results of individuals' tests, without their permission.
- ⌘ However, the implications of test results for other family members are explained, and individuals are encouraged to share results as appropriate.

# The Smith Family

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- ⌘ Mr. Smith developed male breast cancer at age 60, and prostate cancer at age 73.
- ⌘ His mother died from ovarian cancer, and his daughter had early-onset breast cancer.

# Previous Research

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- ⌘ Members of the Smith family have previously participated in family studies with the clinical genetics branch.
- ⌘ The research identified a mutation of the BRCA2 gene in the Smith family.

# Smith Family Test Results

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- ⌘ Two of Mr. Smith's daughters and 2 granddaughters elect to be tested for the BRCA2 mutation.
- ⌘ Testing reveals both daughters (one of whom has breast cancer), and both granddaughters have the same BRCA2 mutation.

# John Smith

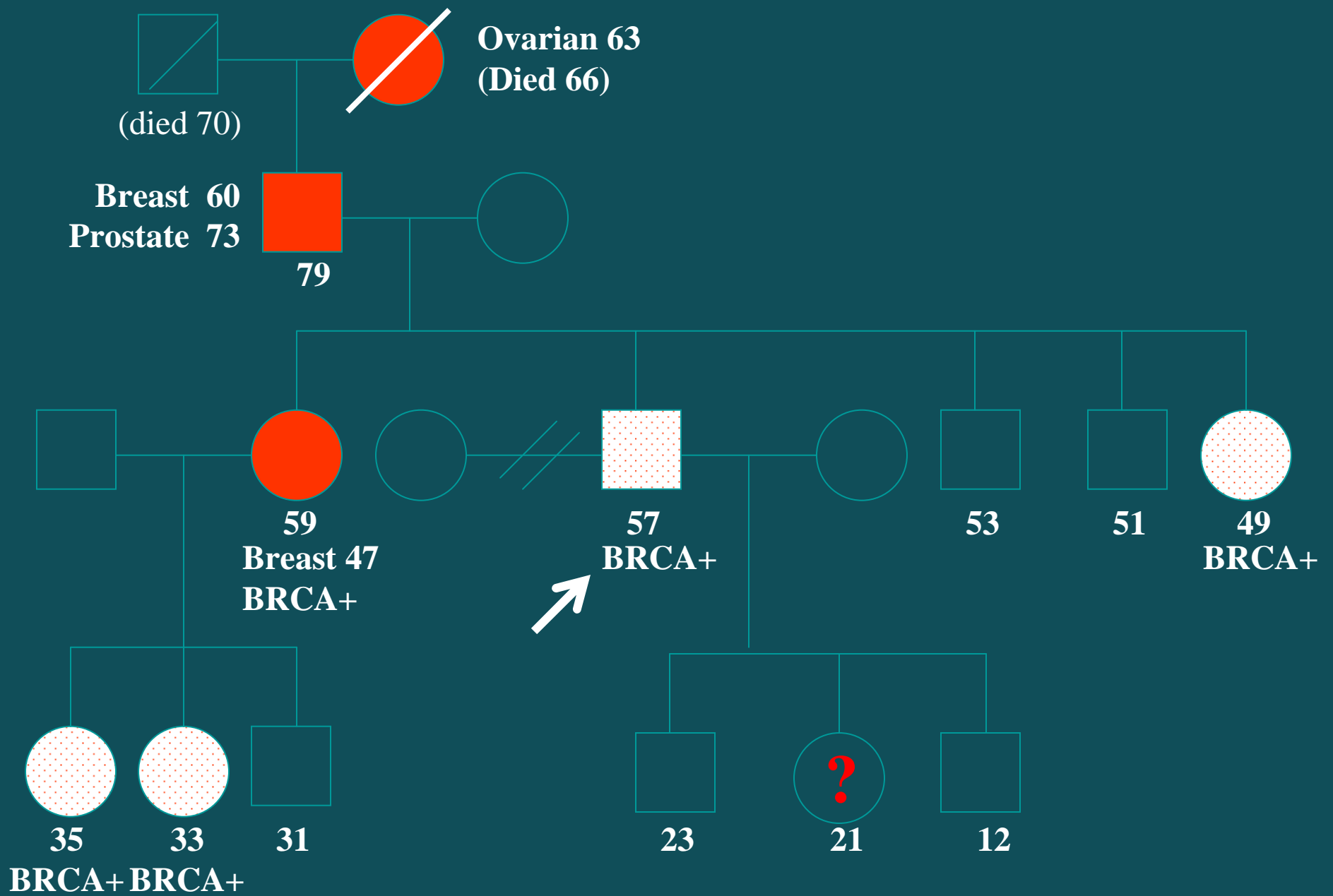
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- ⌘ One of Mr. Smith's sons, John Smith, 57 years old, also elects to be tested for the family's BRCA2 gene mutation.
- ⌘ Before being tested, John reports that he wants to protect his father from feelings of "transmission guilt"

# John Smith's Test Results

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- ⌘ Testing reveals that John Smith has the BRCA2 gene mutation that is related to the risk of breast and ovarian cancer in his family.
- ⌘ The team informs John of his test results.



# John Reports his Results

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- ⌘ With members of the research team present, John tells his family that he does NOT have the BRCA2 gene.
- ⌘ Later, he tells the researchers that he will inform his daughter that he has the gene mutation “when the time is right.”

# Impact on the Children

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- ⌘ The team is worried John will continue to misinform his daughter (and sons), and they will assume they do not have the mutation.
- ⌘ As a result, they may forgo testing and surveillance, even though there is a 50% chance they inherited the mutation related to the family's cancer susceptibility.

# The Team's Dilemma

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- ⌘ The team wonders what, if any, steps it should take.
- ⌘ Is the team obligated to protect John Smith's confidentiality, or is there an ethically acceptable way to warn his children?